

Nabec North American Brain Expression Consortium

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140
papers

19,479
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59
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139
g-index

157
ext. papers

23,569
ext. citations

11.9
avg, IF

5.21
L-index

#	Paper	IF	Citations
140	A hexanucleotide repeat expansion in C9ORF72 is the cause of chromosome 9p21-linked ALS-FTD. <i>Neuron</i> , 2011 , 72, 257-68	13.9	3018
139	Genome-wide association study reveals genetic risk underlying Parkinson's disease. <i>Nature Genetics</i> , 2009 , 41, 1308-12	36.3	1469
138	Large-scale meta-analysis of genome-wide association data identifies six new risk loci for Parkinson's disease. <i>Nature Genetics</i> , 2014 , 46, 989-93	36.3	1261
137	Exome sequencing reveals VCP mutations as a cause of familial ALS. <i>Neuron</i> , 2010 , 68, 857-64	13.9	939
136	Genotype, haplotype and copy-number variation in worldwide human populations. <i>Nature</i> , 2008 , 451, 998-1003	50.4	662
135	Abundant quantitative trait loci exist for DNA methylation and gene expression in human brain. <i>PLoS Genetics</i> , 2010 , 6, e1000952	6	612
134	Common genetic variants influence human subcortical brain structures. <i>Nature</i> , 2015 , 520, 224-9	50.4	601
133	Identification of novel risk loci, causal insights, and heritable risk for Parkinson's disease: a meta-analysis of genome-wide association studies. <i>Lancet Neurology, The</i> , 2019 , 18, 1091-1102	24.1	562
132	A survey of genetic human cortical gene expression. <i>Nature Genetics</i> , 2007 , 39, 1494-9	36.3	413
131	Identification of common variants influencing risk of the tauopathy progressive supranuclear palsy. <i>Nature Genetics</i> , 2011 , 43, 699-705	36.3	386
130	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimer's disease. <i>Nature</i> , 2014 , 505, 550-554	50.4	345
129	A genome-wide association study identifies protein quantitative trait loci (pQTLs). <i>PLoS Genetics</i> , 2008 , 4, e1000072	6	331
128	Genome-wide genotyping in Parkinson's disease and neurologically normal controls: first stage analysis and public release of data. <i>Lancet Neurology, The</i> , 2006 , 5, 911-6	24.1	323
127	Mutations in the Matrin 3 gene cause familial amyotrophic lateral sclerosis. <i>Nature Neuroscience</i> , 2014 , 17, 664-666	25.5	319
126	Distinct DNA methylation changes highly correlated with chronological age in the human brain. <i>Human Molecular Genetics</i> , 2011 , 20, 1164-72	5.6	312
125	Genome-wide Analyses Identify KIF5A as a Novel ALS Gene. <i>Neuron</i> , 2018 , 97, 1268-1283.e6	13.9	296
124	Genome-wide meta-analysis identifies new susceptibility loci for migraine. <i>Nature Genetics</i> , 2013 , 45, 912-917	36.3	276

123	Using exome sequencing to reveal mutations in TREM2 presenting as a frontotemporal dementia-like syndrome without bone involvement. <i>JAMA Neurology</i> , 2013 , 70, 78-84	17.2	257
122	Genome-wide association study of obsessive-compulsive disorder. <i>Molecular Psychiatry</i> , 2013 , 18, 788-98	5.1	244
121	SNCA variants are associated with increased risk for multiple system atrophy. <i>Annals of Neurology</i> , 2009 , 65, 610-4	9.4	232
120	Genetic control of human brain transcript expression in Alzheimer disease. <i>American Journal of Human Genetics</i> , 2009 , 84, 445-58	11	229
119	Loss of VPS13C Function in Autosomal-Recessive Parkinsonism Causes Mitochondrial Dysfunction and Increases PINK1/Parkin-Dependent Mitophagy. <i>American Journal of Human Genetics</i> , 2016 , 98, 500-513	11	225
118	Excessive burden of lysosomal storage disorder gene variants in Parkinson's disease. <i>Brain</i> , 2017 , 140, 3191-3203	11.2	209
117	Chromosome 9p21 in amyotrophic lateral sclerosis in Finland: a genome-wide association study. <i>Lancet Neurology</i> , 2010 , 9, 978-85	24.1	206
116	Genome-wide SNP assay reveals structural genomic variation, extended homozygosity and cell-line induced alterations in normal individuals. <i>Human Molecular Genetics</i> , 2007 , 16, 1-14	5.6	199
115	Novel genetic loci associated with hippocampal volume. <i>Nature Communications</i> , 2017 , 8, 13624	17.4	173
114	Genome-wide genotyping in amyotrophic lateral sclerosis and neurologically normal controls: first stage analysis and public release of data. <i>Lancet Neurology</i> , 2007 , 6, 322-8	24.1	172
113	RNA binding activity of the recessive parkinsonism protein DJ-1 supports involvement in multiple cellular pathways. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008 , 105, 10244-9	11.5	171
112	Genome-wide meta-analysis identifies six novel loci associated with habitual coffee consumption. <i>Molecular Psychiatry</i> , 2015 , 20, 647-656	15.1	167
111	DYT16, a novel young-onset dystonia-parkinsonism disorder: identification of a segregating mutation in the stress-response protein PRKRA. <i>Lancet Neurology</i> , 2008 , 7, 207-15	24.1	159
110	A genome-wide genotyping study in patients with ischaemic stroke: initial analysis and data release. <i>Lancet Neurology</i> , 2007 , 6, 414-20	24.1	154
109	Genome-wide Pleiotropy Between Parkinson Disease and Autoimmune Diseases. <i>JAMA Neurology</i> , 2017 , 74, 780-792	17.2	150
108	Novel genetic loci underlying human intracranial volume identified through genome-wide association. <i>Nature Neuroscience</i> , 2016 , 19, 1569-1582	25.5	147
107	Using genome-wide complex trait analysis to quantify 'missing heritability' in Parkinson's disease. <i>Human Molecular Genetics</i> , 2012 , 21, 4996-5009	5.6	145
106	Diagnosis of Parkinson's disease on the basis of clinical and genetic classification: a population-based modelling study. <i>Lancet Neurology</i> , 2015 , 14, 1002-9	24.1	141

105	Genome-wide association study of Tourette's syndrome. <i>Molecular Psychiatry</i> , 2013 , 18, 721-8	15.1	138
104	Parkinson's disease age at onset genome-wide association study: Defining heritability, genetic loci, and Lysynuclein mechanisms. <i>Movement Disorders</i> , 2019 , 34, 866-875	7	136
103	MAPT expression and splicing is differentially regulated by brain region: relation to genotype and implication for tauopathies. <i>Human Molecular Genetics</i> , 2012 , 21, 4094-103	5.6	134
102	Parkinson's disease in GTP cyclohydrolase 1 mutation carriers. <i>Brain</i> , 2014 , 137, 2480-92	11.2	127
101	Genome-wide screen identifies rs646776 near sortilin as a regulator of progranulin levels in human plasma. <i>American Journal of Human Genetics</i> , 2010 , 87, 890-7	11	110
100	mRNA expression, splicing and editing in the embryonic and adult mouse cerebral cortex. <i>Nature Neuroscience</i> , 2013 , 16, 499-506	25.5	107
99	Mutations in GBA2 cause autosomal-recessive cerebellar ataxia with spasticity. <i>American Journal of Human Genetics</i> , 2013 , 92, 245-51	11	101
98	Integration of GWAS SNPs and tissue specific expression profiling reveal discrete eQTLs for human traits in blood and brain. <i>Neurobiology of Disease</i> , 2012 , 47, 20-8	7.5	100
97	A genome-wide association study of myasthenia gravis. <i>JAMA Neurology</i> , 2015 , 72, 396-404	17.2	99
96	A pathway-based analysis provides additional support for an immune-related genetic susceptibility to Parkinson's disease. <i>Human Molecular Genetics</i> , 2013 , 22, 1039-49	5.6	96
95	A two-stage genome-wide association study of sporadic amyotrophic lateral sclerosis. <i>Human Molecular Genetics</i> , 2009 , 18, 1524-32	5.6	91
94	Exome sequencing reveals riboflavin transporter mutations as a cause of motor neuron disease. <i>Brain</i> , 2012 , 135, 2875-82	11.2	90
93	Mutations in the CHCHD10 gene are a common cause of familial amyotrophic lateral sclerosis. <i>Brain</i> , 2014 , 137, e311	11.2	89
92	Missense variant in TREML2 protects against Alzheimer's disease. <i>Neurobiology of Aging</i> , 2014 , 35, 1510.e119-2684	5.19	84
91	Extended tracts of homozygosity identify novel candidate genes associated with late-onset Alzheimer's disease. <i>Neurogenetics</i> , 2009 , 10, 183-90	3	82
90	Comprehensive analysis of LRRK2 in publicly available Parkinson's disease cases and neurologically normal controls. <i>Human Mutation</i> , 2008 , 29, 485-90	4.7	82
89	Application of genome-wide single nucleotide polymorphism typing: simple association and beyond. <i>PLoS Genetics</i> , 2006 , 2, e150	6	79
88	Shared polygenic risk and causal inferences in amyotrophic lateral sclerosis. <i>Annals of Neurology</i> , 2019 , 85, 470-481	9.4	72

87	Exome sequencing reveals an unexpected genetic cause of disease: NOTCH3 mutation in a Turkish family with Alzheimer's disease. <i>Neurobiology of Aging</i> , 2012 , 33, 1008.e17-23	5.6	72
86	NeuroX, a fast and efficient genotyping platform for investigation of neurodegenerative diseases. <i>Neurobiology of Aging</i> , 2015 , 36, 1605.e7-12	5.6	70
85	Genomewide association study of Parkinson's disease clinical biomarkers in 12 longitudinal patients' cohorts. <i>Movement Disorders</i> , 2019 , 34, 1839-1850	7	69
84	Genetic modifiers of risk and age at onset in GBA associated Parkinson's disease and Lewy body dementia. <i>Brain</i> , 2020 , 143, 234-248	11.2	69
83	Measures of autozygosity in decline: globalization, urbanization, and its implications for medical genetics. <i>PLoS Genetics</i> , 2009 , 5, e1000415	6	63
82	Discovery and functional prioritization of Parkinson's disease candidate genes from large-scale whole exome sequencing. <i>Genome Biology</i> , 2017 , 18, 22	18.3	62
81	Efficacy of Exome-Targeted Capture Sequencing to Detect Mutations in Known Cerebellar Ataxia Genes. <i>JAMA Neurology</i> , 2018 , 75, 591-599	17.2	58
80	NeuroChip, an updated version of the NeuroX genotyping platform to rapidly screen for variants associated with neurological diseases. <i>Neurobiology of Aging</i> , 2017 , 57, 247.e9-247.e13	5.6	54
79	Moving beyond neurons: the role of cell type-specific gene regulation in Parkinson's disease heritability. <i>Npj Parkinsons Disease</i> , 2019 , 5, 6	9.7	51
78	Allelic heterogeneity and more detailed analyses of known loci explain additional phenotypic variation and reveal complex patterns of association. <i>Human Molecular Genetics</i> , 2011 , 20, 4082-92	5.6	51
77	Expanding Parkinson's disease genetics: novel risk loci, genomic context, causal insights and heritable risk		51
76	Frequency of Loss of Function Variants in LRRK2 in Parkinson Disease. <i>JAMA Neurology</i> , 2018 , 75, 1416-1422	17.2	50
75	Genetic comorbidities in Parkinson's disease. <i>Human Molecular Genetics</i> , 2014 , 23, 831-41	5.6	49
74	Exome sequencing in Brown-Vialetto-van Laere syndrome. <i>American Journal of Human Genetics</i> , 2010 , 87, 567-9; author reply 569-70	11	48
73	Mitochondria function associated genes contribute to Parkinson's Disease risk and later age at onset. <i>Npj Parkinsons Disease</i> , 2019 , 5, 8	9.7	47
72	Analysis of genome-wide association studies of Alzheimer disease and of Parkinson disease to determine if these 2 diseases share a common genetic risk. <i>JAMA Neurology</i> , 2013 , 70, 1268-76	17.2	46
71	Genetic variability in CLU and its association with Alzheimer's disease. <i>PLoS ONE</i> , 2010 , 5, e9510	3.7	46
70	Age-associated changes in gene expression in human brain and isolated neurons. <i>Neurobiology of Aging</i> , 2013 , 34, 1199-209	5.6	44

69	Genomewide SNP assay reveals mutations underlying Parkinson disease. <i>Human Mutation</i> , 2008 , 29, 315-22	4.7	44
68	CHCHD2 and Parkinson's disease. <i>Lancet Neurology, The</i> , 2015 , 14, 678-9	24.1	43
67	Imputation of variants from the 1000 Genomes Project modestly improves known associations and can identify low-frequency variant-phenotype associations undetected by HapMap based imputation. <i>PLoS ONE</i> , 2013 , 8, e64343	3.7	42
66	Next-generation sequencing reveals substantial genetic contribution to dementia with Lewy bodies. <i>Neurobiology of Disease</i> , 2016 , 94, 55-62	7.5	41
65	The endocytic membrane trafficking pathway plays a major role in the risk of Parkinson's disease. <i>Movement Disorders</i> , 2019 , 34, 460-468	7	40
64	Additional rare variant analysis in Parkinson's disease cases with and without known pathogenic mutations: evidence for oligogenic inheritance. <i>Human Molecular Genetics</i> , 2016 , 25, 5483-5489	5.6	40
63	Initial assessment of the pathogenic mechanisms of the recently identified Alzheimer risk Loci. <i>Annals of Human Genetics</i> , 2013 , 77, 85-105	2.2	40
62	Resolving the polymorphism-in-probe problem is critical for correct interpretation of expression QTL studies. <i>Nucleic Acids Research</i> , 2013 , 41, e88	20.1	38
61	ABCA7 p.G215S as potential protective factor for Alzheimer's disease. <i>Neurobiology of Aging</i> , 2016 , 46, 235.e1-9	5.6	33
60	A comprehensive analysis of SNCA-related genetic risk in sporadic parkinson disease. <i>Annals of Neurology</i> , 2018 , 84, 117-129	9.4	33
59	Genetics of early-onset Parkinson's disease in Finland: exome sequencing and genome-wide association study. <i>Neurobiology of Aging</i> , 2017 , 53, 195.e7-195.e10	5.6	32
58	Association of tau haplotype-tagging polymorphisms with Parkinson's disease in diverse ethnic Parkinson's disease cohorts. <i>Neurodegenerative Diseases</i> , 2006 , 3, 327-33	2.3	32
57	TBK1 is associated with ALS and ALS-FTD in Sardinian patients. <i>Neurobiology of Aging</i> , 2016 , 43, 180.e1-55.6		32
56	Genome sequencing analysis identifies new loci associated with Lewy body dementia and provides insights into its genetic architecture. <i>Nature Genetics</i> , 2021 , 53, 294-303	36.3	31
55	The Parkinson's Disease Mendelian Randomization Research Portal. <i>Movement Disorders</i> , 2019 , 34, 1864-1872		29
54	Exome sequencing in an SCA14 family demonstrates its utility in diagnosing heterogeneous diseases. <i>Neurology</i> , 2012 , 79, 127-31	6.5	29
53	Exome sequencing identifies a novel TRPV4 mutation in a CMT2C family. <i>Neurology</i> , 2012 , 79, 192-4	6.5	29
52	Use of support vector machines for disease risk prediction in genome-wide association studies: concerns and opportunities. <i>Human Mutation</i> , 2012 , 33, 1708-18	4.7	28

51	Association of a Novel ACTA1 Mutation With a Dominant Progressive Scapulooperoneal Myopathy in an Extended Family. <i>JAMA Neurology</i> , 2015 , 72, 689-98	17.2	27
50	Penetrance of Parkinson's Disease in LRRK2 p.G2019S Carriers Is Modified by a Polygenic Risk Score. <i>Movement Disorders</i> , 2020 , 35, 774-780	7	27
49	Structural genomic variation in ischemic stroke. <i>Neurogenetics</i> , 2008 , 9, 101-8	3	26
48	The Parkinson's Disease Genome-Wide Association Study Locus Browser. <i>Movement Disorders</i> , 2020 , 35, 2056-2067	7	26
47	Mutation analysis of the MS4A and TREM gene clusters in a case-control Alzheimer's disease data set. <i>Neurobiology of Aging</i> , 2016 , 42, 217.e7-217.e13	5.6	24
46	Insufficient evidence for pathogenicity of SNCA His50Gln (H50Q) in Parkinson's disease. <i>Neurobiology of Aging</i> , 2018 , 64, 159.e5-159.e8	5.6	23
45	Mendelian adult-onset leukodystrophy genes in Alzheimer's disease: critical influence of CSF1R and NOTCH3. <i>Neurobiology of Aging</i> , 2018 , 66, 179.e17-179.e29	5.6	23
44	Genetic determinants of survival in progressive supranuclear palsy: a genome-wide association study. <i>Lancet Neurology</i> , 2021 , 20, 107-116	24.1	23
43	Transcriptomic profiling of the human brain reveals that altered synaptic gene expression is associated with chronological aging. <i>Scientific Reports</i> , 2017 , 7, 16890	4.9	22
42	Exome sequencing: an efficient diagnostic tool for complex neurodegenerative disorders. <i>European Journal of Neurology</i> , 2013 , 20, 486-492	6	20
41	Pathogenic Huntingtin Repeat Expansions in Patients with Frontotemporal Dementia and Amyotrophic Lateral Sclerosis. <i>Neuron</i> , 2021 , 109, 448-460.e4	13.9	20
40	Large-scale pathway specific polygenic risk and transcriptomic community network analysis identifies novel functional pathways in Parkinson disease. <i>Acta Neuropathologica</i> , 2020 , 140, 341-358	14.3	19
39	Clinical and genetic analyses of familial and sporadic frontotemporal dementia patients in Southern Italy. <i>Alzheimers and Dementia</i> , 2017 , 13, 858-869	1.2	18
38	The Genetic Architecture of Parkinson Disease in Spain: Characterizing Population-Specific Risk, Differential Haplotype Structures, and Providing Etiologic Insight. <i>Movement Disorders</i> , 2019 , 34, 1851-1863	7	18
37	Identification of Candidate Parkinson Disease Genes by Integrating Genome-Wide Association Study, Expression, and Epigenetic Data Sets. <i>JAMA Neurology</i> , 2021 , 78, 464-472	17.2	17
36	SLC25A46 Mutations Associated with Autosomal Recessive Cerebellar Ataxia in North African Families. <i>Neurodegenerative Diseases</i> , 2017 , 17, 208-212	2.3	16
35	Mutation Frequency of the Major Frontotemporal Dementia Genes, MAPT, GRN and C9ORF72 in a Turkish Cohort of Dementia Patients. <i>PLoS ONE</i> , 2016 , 11, e0162592	3.7	16
34	SNCA and mTOR Pathway Single Nucleotide Polymorphisms Interact to Modulate the Age at Onset of Parkinson's Disease. <i>Movement Disorders</i> , 2019 , 34, 1333-1344	7	14

33	Comprehensive promoter level expression quantitative trait loci analysis of the human frontal lobe. <i>Genome Medicine</i> , 2016 , 8, 65	14.4	14
32	Establishing the role of rare coding variants in known Parkinson's disease risk loci. <i>Neurobiology of Aging</i> , 2017 , 59, 220.e11-220.e18	5.6	11
31	ADORA1 mutations are not a common cause of Parkinson's disease and dementia with Lewy bodies. <i>Movement Disorders</i> , 2017 , 32, 298-299	7	10
30	Whole-genome sequencing to understand the genetic architecture of common gene expression and biomarker phenotypes. <i>Human Molecular Genetics</i> , 2015 , 24, 1504-12	5.6	7
29	Regulatory sites for splicing in human basal ganglia are enriched for disease-relevant information. <i>Nature Communications</i> , 2020 , 11, 1041	17.4	6
28	A 7.5-Mb duplication at chromosome 11q21-11q22.3 is associated with a novel spastic ataxia syndrome. <i>Movement Disorders</i> , 2015 , 30, 262-6	7	6
27	Parkinson disease age of onset GWAS: defining heritability, genetic loci and a-synuclein mechanisms		6
26	Investigation of Autosomal Genetic Sex Differences in Parkinson's Disease. <i>Annals of Neurology</i> , 2021 , 90, 35-42	9.4	6
25	Mutational Analysis of Known ALS Genes in an Italian Population-Based Cohort. <i>Neurology</i> , 2021 , 96, e600-e609	6.5	6
24	Exome sequencing establishes a gelsolin mutation as the cause of inherited bulbar-onset neuropathy. <i>Muscle and Nerve</i> , 2017 , 56, 1001-1005	3.4	5
23	Accelerating Medicines Partnership: Parkinson's Disease. Genetic Resource. <i>Movement Disorders</i> , 2021 , 36, 1795-1804	7	5
22	Association of Variants in the SPTLC1 Gene With Juvenile Amyotrophic Lateral Sclerosis. <i>JAMA Neurology</i> , 2021 , 78, 1236-1248	17.2	5
21	Age-modulated association between prefrontal NAA and the BDNF gene. <i>International Journal of Neuropsychopharmacology</i> , 2013 , 16, 1185-93	5.8	4
20	Exome Sequencing Reveals VCP Mutations as a Cause of Familial ALS. <i>Neuron</i> , 2011 , 69, 397	13.9	4
19	Association of a Common Genetic Variant with Parkinson's Disease is Propagated through Microglia		4
18	The Parkinson's Disease Mendelian Randomization Research Portal		3
17	Penetrance of Parkinson's disease in LRRK2 p.G2019S carriers is modified by a polygenic risk score		3
16	A SINE-VNTR- in the LRIG2 Promoter Is Associated with Gene Expression at the Locus. <i>International Journal of Molecular Sciences</i> , 2020 , 21,	6.3	3

15	Identification of sixteen novel candidate genes for late onset Parkinson's disease. <i>Molecular Neurodegeneration</i> , 2021 , 16, 35	19	3
14	The Parkinson's Disease DNA Variant Browser. <i>Movement Disorders</i> , 2021 , 36, 1250-1258	7	3
13	Another locus, a new method. <i>Brain</i> , 2010 , 133, 3492-3	11.2	2
12	RNA sequencing of whole blood reveals early alterations in immune cells and gene expression in Parkinson's disease. <i>Nature Aging</i> , 2021 , 1, 734-747		2
11	Genome-Wide Analyses Identify KIF5A as a Novel ALS Gene. <i>SSRN Electronic Journal</i> ,	1	2
10	Accelerating Medicines Partnership: Parkinson's Disease. Genetic Resource		2
9	Exploring dementia and neuronal ceroid lipofuscinosis genes in 100 FTD-like patients from 6 towns and rural villages on the Adriatic Sea coast of Apulia. <i>Scientific Reports</i> , 2021 , 11, 6353	4.9	2
8	Shared Genetics of Multiple System Atrophy and Inflammatory Bowel Disease. <i>Movement Disorders</i> , 2021 , 36, 449-459	7	2
7	Quality Control Metrics for Whole Blood Transcriptome Analysis in the Parkinson's Progression Markers Initiative (PPMI)		2
6	Finding genetically-supported drug targets for Parkinson's disease using Mendelian randomization of the druggable genome.. <i>Nature Communications</i> , 2021 , 12, 7342	17.4	2
5	MIDN locus structural variants and Parkinson's Disease risk. <i>Annals of Clinical and Translational Neurology</i> , 2020 , 7, 602-603	5.3	1
4	A simple and efficient algorithm for genome-wide homozygosity analysis in disease. <i>Molecular Systems Biology</i> , 2009 , 5, 304	12.2	1
3	The genetic architecture of Parkinson disease in Spain: characterizing population-specific risk, differential haplotype structures, and providing etiologic insight		1
2	The Parkinson's Disease GWAS Locus Browser		1
1	Replication assessment of NUS1 variants in Parkinson's disease. <i>Neurobiology of Aging</i> , 2021 , 101, 300.e15-300.e3	5.3	1